



## HFE2 gene

hemochromatosis type 2 (juvenile)

### Normal Function

The *HFE2* gene provides instructions for making a protein called hemojuvelin. This protein is made in the liver, heart, and muscles used for movement (skeletal muscles). Researchers recently discovered that hemojuvelin plays a role in maintaining iron balance in the body. Although its exact function is unclear, hemojuvelin appears to regulate the levels of another protein called hepcidin. Hepcidin also plays a key role in maintaining proper iron levels in the body.

### Health Conditions Related to Genetic Changes

#### hereditary hemochromatosis

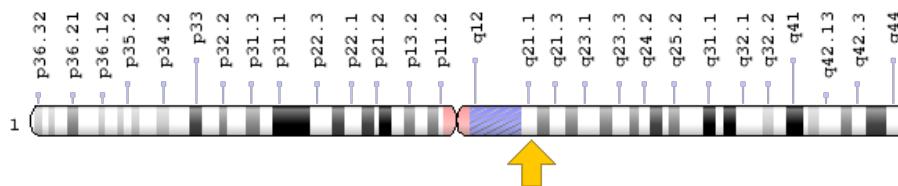
Researchers have identified more than 20 *HFE2* gene mutations that cause type 2 hemochromatosis, a form of hereditary hemochromatosis that begins during childhood or adolescence. Most *HFE2* gene mutations change one of the protein building blocks (amino acids) used to make hemojuvelin. Most frequently, the amino acid glycine is replaced by the amino acid valine at protein position 320 (written as Gly320Val). Other mutations create a premature stop signal in the instructions for making the hemojuvelin protein. As a result, an abnormally small protein is made.

Mutations in the *HFE2* gene lead to an altered hemojuvelin protein that cannot function properly. Without adequate hemojuvelin, levels of the protein hepcidin are reduced and iron balance is disturbed. As a result, too much iron is absorbed during digestion, which leads to iron overload and damage to tissues and organs in the body.

## Chromosomal Location

Cytogenetic Location: 1q21.1, which is the long (q) arm of chromosome 1 at position 21.1

Molecular Location: base pairs 146,017,468 to 146,021,822 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- HFE2A
- HJV
- JH
- RGM domain family, member C
- RGMC
- RGMC\_HUMAN

## Additional Information & Resources

### GeneReviews

- Juvenile Hereditary Hemochromatosis  
<https://www.ncbi.nlm.nih.gov/books/NBK1170>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HFE2%5BTIAB%5D%29+OR+%28hemochromatosis+type+2%5BTIAB%5D%29+OR+%28hemochromatosis+AND+1q%5BTIAB%5D%29%29+OR+%28%28hemojuvelin%5BTIAB%5D%29+OR+%28HFE2A%5BTIAB%5D%29+OR+%28HJV%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- HEMOJUVELIN  
<http://omim.org/entry/608374>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_HFE2.html](http://atlasgeneticsoncology.org/Genes/GC_HFE2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=HFE2%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=4887](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4887)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/148738>
- UniProt  
<http://www.uniprot.org/uniprot/Q6ZVN8>

## **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/HFE2>

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